CONGENITAL ADRENAL HYPERPLASIA (CAH)

(con–JEN-I-tal ah DREE-nal HIGH-per-PLAY-ze-an)

What is it?

Congenital adrenal hyperplasia (CAH) is a term used to describe a group of distinct inherited disorders (*autosomal recessive inheritance*), each with a deficiency in secretion of cortisol, and in some instances, also a deficiency in secretion of the salt-retaining hormone, aldosterone. CAH is the most common cause of ambiguous genitalia in females, and can cause acute life-threatening adrenal crisis in both males and females in the neonatal period. The most common form of CAH (more than 90% of cases) is caused by deficiency of the adrenal enzyme, *21-hydroxylase* (21-OH). Depending on the extent of the enzyme deficiency, the disorder presents as either a salt-losing form (75% of cases), or a non-salt-losing form (25% of cases). Many studies have shown that newborn screening for CAH results in earlier diagnosis and treatment, often prior to the development of adrenal crisis. Infants with symptoms of impending adrenal crisis include vomiting, dehydration, weight loss, poor feeding, electrolyte imbalance and lethargy.

How do you get it?

The main problem in CAH is the inability of the adrenal glands to produce enough cortisol in the non-salt wasting form, or enough cortisol and salt-retaining hormone in the salt-wasting form. This inability of the body to produce these hormones is the reason newborns and children who do not receive treatment become very sick with this disorder. Symptoms of salt wasting CAH include frequent urination, and in some cases, poor feeding, which can rapidly progress to vomiting, dehydration, electrolyte changes and cardiac arrhythmia. Infants with CAH who are not diagnosed and treated early are particularly susceptible to sudden death in the first weeks of life. Instead of making cortisol, the hormonal raw materials, which usually make cortisol, are shifted away to make other hormones, specifically male sexual hormones (androgens). As a result, more androgens are produced than necessary.

Female infants with 21 hydroxylase deficient CAH usually have some degree of virilization (ambiguous genitalia) due to their exposure to excessive androgen levels in utero. Although male infants usually appear normal at birth, they may have an enlarged penis and scrotum with increased pigmentation. Symptoms of salt wasting CAH include frequent urination, and in some cases, poor feeding, which can rapidly progress to vomiting, dehydration, electrolyte changes and cardiac arrhythmia. Infants with CAH who are not diagnosed and treated early are particularly susceptible to

sudden death in the first few weeks of life. In older children, CAH may result in rapid growth and precocious puberty with premature skeletal maturation.

How common is it?

Congenital adrenal hyperplasia occurs in 1 out of every 13,000 births in Missouri.

The vast majority (90 percent) of CAH cases result from 21-hydroxylase deficiency. The only form of CAH detected by newborn screening, 21-hydroxylase deficiency is inherited in an autosomal recessive pattern. As with other autosomal recessive disorders, the parents of a child with CAH are unaffected, healthy carriers of the condition have one normal gene and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene, resulting in CAH. Carrier parents have a 50 percent chance of having a child who is an unaffected carrier and a 25 percent chance of having an unaffected, non-carrier child. These risks hold true for each pregnancy. All siblings of infants diagnosed with congenital adrenal hyperplasia should be tested; genetic counseling services should be offered to the family.

How is it treated?

Treatment for CAH includes lifetime daily medication. Oral hydrocortisone in children and prednisone or dexamethasone for older individuals, replaces missing cortisol. Hydrocortisone is usually given at regular intervals three times a day. In cases of salt wasting CAH, in addition to hydrocortisone, fludrocortisone is prescribed to correct aldosterone deficiency. Infants and small children with salt wasting CAH also may require salt tablets as a dietary supplement. Regulation of medication dosage is vital, as improper dosage can result in either growth delay or premature bone epiphyseal closure. Female infants with ambiguous genitalia may require re-constructive surgery.

If your child needs additional testing or diagnostic evaluation, it is important that you follow through with the pediatrician's and/or specialist's recommendations for additional testing and referrals.

Treatment is life long and compliance with medication and frequent blood monitoring are imperative to the child's health, growth and development. Although children with CAH are usually healthy, any illness (for example, fever, vomiting or injury) requires prompt notification of the child's physician, as the cortisol dosage may need to be increased. In addition, parents should keep injectable hydrocortisone on hand at all times. If the

child has repeated vomiting or is unable to hold down fluids, parents should call the specialist immediately. In emergencies, parents must be prepared to administer injectable hydrocortisone if instructed to do so by the physician. Children and adolescents with CAH should wear medical identification bracelets or necklaces to alert health care providers to his/her condition and to insure proper medication is provided in an emergency

Treatment is not curative and all morbidity cannot necessarily be prevented. Long-term management, monitoring and compliance with treatment recommendations are essential to the child's well being. A multidisciplinary approach is recommended and should include the following specialties: pediatrics, endocrinology and, in some cases, pediatric reconstructive surgery. Infants and children with congenital adrenal hyperplasia should have regular follow-up appointments with a pediatric endocrinologist to regulate medication regimens.

Where can I get services?

Provision of the names below does not necessarily include all hospitals or private practice physicians who may treat children with CAH.

Cardinal Glennon Memorial Hospital for Children
St. Louis, MO
314-577-5648

Children's Mercy Hospital
Kansas City, MO
816-234-3804

St. Louis Children's Hospital
St. Louis, MO
314-454-6051

University Hospital and Clinics
Columbia, MO
573-882-6979

What does DHSS offer?

State Public Health Laboratory – Newborn Screen

Related Links

Congenital Adrenal hyperplasia Research Education and Support Foundation http://www.caresfoundation.org

Medline Plus (National Library of Medicine and the National Institutes of Health) www.medlineplus.gov

National Institutes of Health www.nih.gov

National Newborn Screening and Genetics Resource Center,

http://genes-r-us.uthscsa.edu.

Organization for Endocrine and Metabolic Disorders
www.niddk.nih.gov

GeneTests http://www.genetests.org

The MAGIC Foundation http://www.magicfoundation.org